

Abstract: The present invention describes a method for the analysis of methylation patterns comprising the following steps: a) isolation of genomic nucleic acids from a biological sample, b) amplification of one or more target nucleic acids of said genomic nucleic acids in a manner whereby the methylation patterns of said genomic nucleic acids are maintained in the amplificate nucleic acid, c) performing mass spectrometry on said amplified nucleic acid or fragments thereof to obtain mass spectra; d) evaluating the obtained mass spectra and e) determining the methylation pattern and/or methylation status of the sample. The disclosed invention provides novel methods for the analysis of cytosine methylation patterns within genomic DNA samples. Said method comprises a methylation retaining enzymatic amplification of a test nucleic acid sample, followed by mass spectrometric analysis of the amplificate nucleic acids.

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